Carnitine Uptake Defect (CUD)*

A fatty acid oxidation disorder

What is it?

Carnitine Uptake Defect (also known as CUD) is an inherited fatty acid oxidation disorder. Patients with fatty acid oxidation disorders, like CUD, cannot breakdown fats (long chain fatty acids) to energy. This is because the breakdown of fats in the body relies on adequate levels of Carnitine. Once the body uses up its primary source of energy (glucose, or blood sugar), the body begins to fail because it cannot then make energy from fats because the Carnitine level is too low. Therefore, people with CUD must take a Carnitine supplement and may benefit from frequent feedings and avoidance of fasting.

What are the symptoms?

A person with CUD can appear normal at birth. Symptoms such as vomiting, increased tiredness, behavior changes and hypoglycemia can occur. A person with CUD may also develop enlargement of the heart or heat failure, muscle weakness, liver problems, coma or sudden unexpected death. Many symptoms of CUD can be prevented by immediate treatment and lifelong management. People with CUD typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

CUD is inherited in an autosomal recessive manner. This means that for a person to be affected with CUD, he or she must have inherited two non-working copies of the gene responsible for causing CUD. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have CUD. Typically, there is no family history of CUD in an affected person. However, some carriers of CUD may have some signs and symptoms related to this condition. The newborns mother should be tested as well because several cases of maternal CUD have been identified following an abnormal newborn screening result in their off spring. CUD is a rare fatty acid oxidation disorder; the total number of people affected with CUD is not known.

How is it detected?

CUD may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

CUD is treated by supplementation with a medication called carnitine. Sometimes, a low fat, high carbohydrate diet, frequent feeding diet regimen has been used.

***There is a lower probability of detection of this disorder during the immediate newborn period.**

For more information:

Genetics Home Reference
Website: http://www.ghr.nlm.nih.gov

Save Babies Through Screening Foundation
4 Manor View Circle
Malvern, PA 19355-1622
Toll Free Phone: 1-888-454-3383
Fax: (610) 993-0545
Email: email@savebabies.org
Website: http://www.savebabies.org

FOD (Fatty Oxidation Disorder) Family Support Group
1559 New Garden Rd, 2E
Greensboro, NC 27410
Phone: (336) 547-8682 [8am - 8pm EST every day]
Fax: (336) 292-0536 [email/call ahead between 8am and 8pm before faxing]
Email: deb@fodsupport.org
Website: http://www.fodsupport.org

United Mitochondrial Disease Foundation
8085 Saltsburg Road, Suite 201
Pittsburgh, PA 15239
Phone: (412) 793-8077
FAX: (412) 793-6477
email: info@umdf.org
website: http://www.umdf.org